

Modern biomedical research

is increasingly supported by

information technologies. Biolo-

gists and physicians rely not only

on the biomedical literature (e.g.,

MEDLINE), but also on the

many knowledge bases available

online. While these resources are

undeniably valuable to humans,

most of them are text-based and heterogeneous, and cannot be

easily processed by computers.

The *Biomedical Knowledge Repository* under development at

the National Library of Medicine

addresses these limitations. It

can be understood as a special-

ized version of the Semantic

Web. It consists of an extensive

collection of assertions repre-

sented in a common format.

Logical reasoners extend the

capabilities of the repository by

inferring new knowledge.

Integrating Biomedical Knowledge Experience with Entrez Gene

BENCH TO BEDSIDE

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has_associated_disease

APP → Alzheimer disease

subject predicate object

APP Alzheimer disease

has_associated_disease

PARK1 Parkinson disease

Example of RDF triple

Example of RDF graph combining triples from Entrez Gene and MeSH

Transformation rules (XSLT) Phenotypes has associated disease **XML RDF** RDF JAXP Jena (file) (file) (Oracle) 124 element tags 106 properties 410M triples 2M genes

Converting Entrez Gene to RDF using an XSLT approach



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We converted NCBI's gene information resource *Entrez Gene* from its XML format to RDF, the *Resource Description Framework*. This transformation is not simply syntactic, but also semantic and was achieved through rules created manually and expressed in an XSLT (*Extensible Stylesheet Language Transformation*).



Once converted to RDF, Entrez Gene forms a graph and can be integrated seamlessly with other resources such as the *Medical Subject Headings* (MeSH). Hierarchies in MeSH can then be exploited to query genes, enabling researchers to formulate queries such as "Find all genes involved with neuro-degenerative diseases".

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